



## SLC2A1 gene

solute carrier family 2 member 1

### Normal Function

The *SLC2A1* gene provides instructions for producing a protein called the glucose transporter protein type 1 (GLUT1). The GLUT1 protein is embedded in the outer membrane surrounding cells, where it transports a simple sugar called glucose into cells from the blood or from other cells for use as fuel.

In the brain, the GLUT1 protein is involved in moving glucose, which is the brain's main energy source, across the blood-brain barrier. The blood-brain barrier acts as a boundary between tiny blood vessels (capillaries) and the surrounding brain tissue; it protects the brain's delicate nerve tissue by preventing many other types of molecules from entering the brain. The GLUT1 protein also moves glucose between cells in the brain called glia, which protect and maintain nerve cells (neurons).

### Health Conditions Related to Genetic Changes

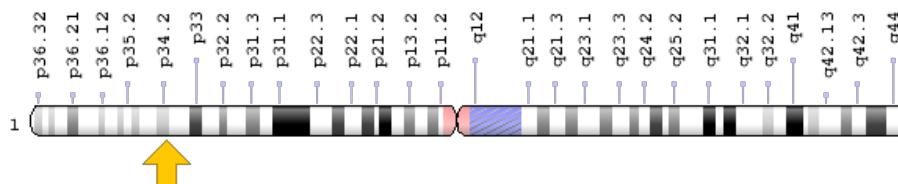
#### GLUT1 deficiency syndrome

More than 150 *SLC2A1* gene mutations have been reported in people with GLUT1 deficiency syndrome. This disorder leads to a variety of neurological symptoms that can include developmental delay, intellectual disability, movement problems, and frequent seizures (epilepsy). The mutations that cause GLUT1 deficiency syndrome reduce or eliminate the function of the GLUT1 protein. Having less functional GLUT1 protein reduces the amount of glucose available to brain cells, which affects brain development and function.

## Chromosomal Location

Cytogenetic Location: 1p34.2, which is the short (p) arm of chromosome 1 at position 34.2

Molecular Location: base pairs 42,925,375 to 42,959,176 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DYT9
- DYT17
- DYT18
- GLUT
- GLUT1
- GTR1\_HUMAN
- MGC141895
- MGC141896
- solute carrier family 2 (facilitated glucose transporter), member 1

## Additional Information & Resources

### Educational Resources

- Molecular Cell Biology (fourth edition, 2000): GLUT1 Transports Glucose into Most Mammalian Cells  
<https://www.ncbi.nlm.nih.gov/books/NBK21669/#A4044>

### GeneReviews

- Glucose Transporter Type 1 Deficiency Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1430>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC2A1%5BTIAB%5D%29+OR+%28%28GLUT%5BTIAB%5D%29+OR+%28GLUT1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

## OMIM

- SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1  
<http://omim.org/entry/138140>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SLC2A1.html](http://atlasgeneticsoncology.org/Genes/GC_SLC2A1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC2A1%5Bgene%5D>
- HGNC Gene Family: Solute carriers  
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=11005](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11005)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6513>
- UniProt  
<http://www.uniprot.org/uniprot/P11166>

## **Sources for This Summary**

- Brockmann K. The expanding phenotype of GLUT1-deficiency syndrome. *Brain Dev.* 2009 Aug; 31(7):545-52. doi: 10.1016/j.braindev.2009.02.008. Epub 2009 Mar 21. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19304421>
- Guo X, Geng M, Du G. Glucose transporter 1, distribution in the brain and in neural disorders: its relationship with transport of neuroactive drugs through the blood-brain barrier. *Biochem Genet.* 2005 Apr;43(3-4):175-87. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15932065>
- Klepper J. Impaired glucose transport into the brain: the expanding spectrum of glucose transporter type 1 deficiency syndrome. *Curr Opin Neurol.* 2004 Apr;17(2):193-6. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15021248>

- Leen WG, Klepper J, Verbeek MM, Leferink M, Hofste T, van Engelen BG, Wevers RA, Arthur T, Bahi-Buisson N, Ballhausen D, Bekhof J, van Bogaert P, Carrilho I, Chabrol B, Champion MP, Coldwell J, Clayton P, Donner E, Evangelou A, Ebinger F, Farrell K, Forsyth RJ, de Goede CG, Gross S, Grunewald S, Holthausen H, Jayawant S, Lachlan K, Laugel V, Leppig K, Lim MJ, Mancini G, Marina AD, Martorell L, McMenamin J, Meuwissen ME, Mundy H, Nilsson NO, Panzer A, Poll-The BT, Rauscher C, Rouselle CM, Sandvig I, Scheffner T, Sheridan E, Simpson N, Sykora P, Tomlinson R, Trounce J, Webb D, Weschke B, Scheffer H, Willemsen MA. Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. *Brain*. 2010 Mar;133(Pt 3):655-70. doi: 10.1093/brain/awp336. Epub 2010 Feb 2.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20129935>
- Pascual JM, Wang D, Hinton V, Engelstad K, Saxena CM, Van Heertum RL, De Vivo DC. Brain glucose supply and the syndrome of infantile neuroglycopenia. *Arch Neurol*. 2007 Apr;64(4):507-13. Epub 2007 Feb 12.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17296829>
- Pascual JM, Wang D, Lecumberri B, Yang H, Mao X, Yang R, De Vivo DC. GLUT1 deficiency and other glucose transporter diseases. *Eur J Endocrinol*. 2004 May;150(5):627-33. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15132717>
- Pearson TS, Akman C, Hinton VJ, Engelstad K, De Vivo DC. Phenotypic spectrum of glucose transporter type 1 deficiency syndrome (Glut1 DS). *Curr Neurol Neurosci Rep*. 2013 Apr;13(4):342. doi: 10.1007/s11910-013-0342-7. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23443458>
- Rotstein M, Engelstad K, Yang H, Wang D, Levy B, Chung WK, De Vivo DC. Glut1 deficiency: inheritance pattern determined by haploinsufficiency. *Ann Neurol*. 2010 Dec;68(6):955-8. doi: 10.1002/ana.22088.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20687207>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2994988/>
- OMIM: SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1  
<http://omim.org/entry/138140>
- Simpson IA, Carruthers A, Vannucci SJ. Supply and demand in cerebral energy metabolism: the role of nutrient transporters. *J Cereb Blood Flow Metab*. 2007 Nov;27(11):1766-91. Epub 2007 Jun 20. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17579656>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2094104/>
- Wang D, Pascual JM, Yang H, Engelstad K, Jhung S, Sun RP, De Vivo DC. Glut-1 deficiency syndrome: clinical, genetic, and therapeutic aspects. *Ann Neurol*. 2005 Jan;57(1):111-8.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15622525>

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